



## SOX2 gene

SRY-box 2

### Normal Function

The SOX2 gene provides instructions for making a protein that plays a critical role in the formation of many different tissues and organs during embryonic development. The SOX2 protein is especially important for the development of the eyes. This protein regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, the SOX2 protein is called a transcription factor.

### Health Conditions Related to Genetic Changes

combined pituitary hormone deficiency

microphthalmia

septo-optic dysplasia

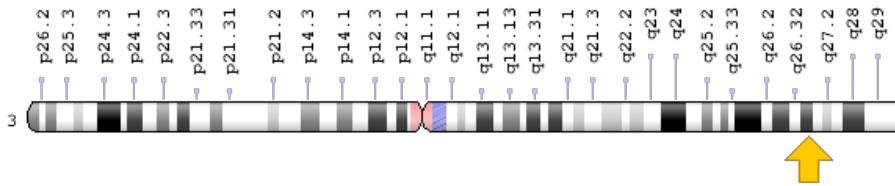
SOX2 anophthalmia syndrome

At least 33 mutations in the SOX2 gene have been found to cause SOX2 anophthalmia syndrome. Some of these mutations prevent the gene from making any SOX2 protein, while others result in the production of an abnormally short, nonfunctional version of the protein. A few mutations change single protein building blocks (amino acids) in the SOX2 protein. All of these mutations disrupt the protein's ability to regulate genes essential for normal development of the eyes and other parts of the body. Abnormal development of these structures causes the signs and symptoms of SOX2 anophthalmia syndrome.

## Chromosomal Location

Cytogenetic Location: 3q26.33, which is the long (q) arm of chromosome 3 at position 26.33

Molecular Location: base pairs 181,711,924 to 181,714,436 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ANOP3
- MCOPS3
- MGC2413
- sex-determining region Y-box 2
- SOX2\_HUMAN
- SRY (sex determining region Y)-box 2
- SRY box 2
- SRY-related HMG-box gene 2
- transcription factor SOX2

## Additional Information & Resources

### Educational Resources

- Developmental Biology (sixth edition, 2000): Transcription Factors  
<https://www.ncbi.nlm.nih.gov/books/NBK10023/#A763>

### GeneReviews

- Microphthalmia/Anophthalmia/Coloboma Spectrum  
<https://www.ncbi.nlm.nih.gov/books/NBK1378>
- SOX2-Related Eye Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1300>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SOX2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28stem+cell%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

## OMIM

- SRY-BOX 2  
<http://omim.org/entry/184429>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/SOX2ID44064ch3q26.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SOX2%5Bgene%5D>
- HGNC Gene Family: SRY-boxes  
<http://www.genenames.org/cgi-bin/genefamilies/set/757>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=11195](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11195)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6657>
- UniProt  
<http://www.uniprot.org/uniprot/P48431>

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